

Table 3. XK Mutations

Exon	DNA Change	Protein Change	Reference
Whole gene	Major deletion	Absence of XK protein	Ho et al 1994; Danek, Rubio et al 2001; Danek, Tison et al 2001; El Nemer et al 2000; Peng et al 2007
Promotor + exon 1	Major deletion	Absence of XK protein	Ho et al 1994
Promotor + exon 1	Major deletion	Absence of XK protein	Wendel et al 2004
1	Major deletion	Absence of XK protein	Danek, Rubio et al 2001
Exons 1 + 2	Major deletion	Absence of XK protein	Walker et al 2006
1	254delG	V58YfsX71	Zeman et al 2005
1	189G>A	W36X	Malandrini et al 1994; Danek, Rubio et al 2001
IVS1	IVS1+1G>C	Splice-site mutation	Russo et al 2002
2	7453 bp Major deletion	Unknown	Singleton et al 2003
2	350delT ¹	Y90TfsX39	Ho et al 1996
2	479C>T	R133X	Dotti et al 2000; Danek, Rubio et al 2001
2	533_534insC	Q151PfsX47	Ueyama et al 2000, Starling et al 2005
2	545C>T	Q155X	Danek, Rubio et al 2001
IVS2	IVS2+1G>A ²	Splice-site mutation	Swash et al 1983, Ho et al 1994
IVS2	IVS2+5G>A	Splice-site mutation	Daniels et al 1996
IVS2	IVS2-1G>A ³	Splice-site mutation	Swash et al 1983, Ho et al 1994
3	746C>G	R222G	Russo et al 2002
3	768_769delTT	F229YfsX35	Danek, Rubio et al 2001

Exon	DNA Change	Protein Change	Reference
3	789G>A	W236X	Danek, Rubio et al 2001
3	853delG	W257CfsX10	Danek, Rubio et al 2001
3	938_942delCTCTA	L286YfsX15	Danek, Rubio et al 2001
3	962T>C ⁴	C294R	Danek, Rubio et al 2001
3	977C>T ⁵	Q299X	Jung et al 2001
3	1020_1033del ⁶	N313TfsX23	Allen et al 1961; Danek, Rubio et al 2001
3	1023G>A ⁷	W314X	Supple et al 2001
3	1061G>A	E327K	Jung et al 2003
3	1095delT ⁸	F338SfsX70	Hanaoka et al 1999

Notes:

- A. The numbering of the cDNA starts from the first base in the GenBank entry Z32684, i.e. the A of the initiation codon is assigned as base 83.
 - B. For the frameshift mutation, the following nomenclature was used: the first amino acid change_fsX_the length of the shifted reading frame ({ HYPERLINK "http://www.genomic.unimelb.edu.au mdi/mutnomen/recs.html" \l "protein" \t "_top" }).
1. OMIM allelic variant 314850.003
 2. OMIM allelic variant 314850.001
 3. OMIM allelic variant 314850.002
 4. OMIM allelic variant 314850.005
 5. OMIM allelic variant 314850.008
 6. OMIM allelic variant 314850.006
 7. OMIM allelic variant 314850.007
 8. OMIM allelic variant 314850.004